Marburg's Disease and Neuromyelitis Optica in Brother/Sister

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Introduction

Marburg's disease (acute MS)

- Rare variant of MS
- Monophasic, progressive course
- Multifocal neurological disturbances
- Rapid deterioration
- High mortality rate

Neuromyelitis optica (NMO)

- Distinct demyelinating entity
- Selective involvement of the optic nerves and spinal cord
- Rare involvement of the brain
- Extensive involvement of the spinal cord; more than 3 vertebral segments
- Progressive or relapsing course
- Immunosuppressive therapy

Methods:

Patient #1

38 year old man:

- No PMH
- Progressive right arm weakness
- One month later: LE weakness with rapid progression
- Four months later: wheelchair bound
- Brain MRI and LP: demyelination
- Eighteen months later: patient expired

Patient #2

Nine years later, patient's younger sister at 43 y.o.:

- ATM treated with IV steroids and recovered
- In the following two years, two more episodes of ATM and one episode of left ON occured
- MRI of the brain with and without gadolinium: unremarkable
- MRI of the spine: extensive demyelination at the spinal cord
- LP: no oligoclonal bands
- NMO-IgG: positive
- On immunosuppressive therapy
- Has moderate disability

Neuroimaging:

Fig 1. T2-weighted sequences, axial. Extensive white matter disease in the brain stem

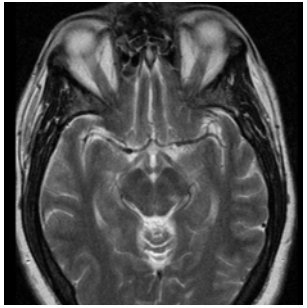
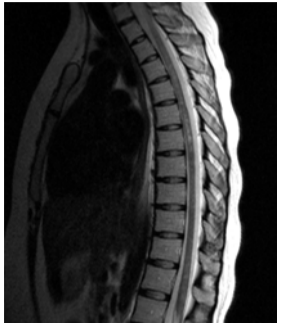


Fig 2. T2-weighted sequences, sag. Signal changes in the thoracic cord.



Conclusions/Discussion:

Familial cases of multiple sclerosis between siblings or parents and children have been reported. A case of NMO in a mother and daughter and another case of NMO and MS between sisters have been documented.

This is the first case report demonstrating the occurrence of two rare demyelinating diseases in a brother and sister. This rare occurrence suggests a complex genetic etiology and probable genetic heterogeneity even between members of the same family. It requires further testing and consideration.

Significance to MS:

Genetic risk in MS is low, but real. It calls for further investigation.

References:

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